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The Meckel Syndrome: Report of Two Japanese Sibs and a Review of Literature

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Two Japanese sibs with the Meckel syndrome are reported. Both babies showed the classical triad of this condition: occipital encephalocele, cystic kidneys, and postaxial polydactyly of all four limbs. The diagnostic criteria and differential diagnosis were reviewed. © 1996 Wiley-Liss, Inc.

KEY WORDS: Meckel syndrome, autosomal recessive inheritance, encephalocele, cystic kidneys, polydactyly

INTRODUCTION

The Meckel syndrome (MS) is a rare autosomal recessive condition showing the triad of occipital encephalocele, cystic kidneys, and postaxial polydactyly of all four limbs.

MS has been reported in families originating from most parts of the world [Salonen, 1984].

The purpose of this paper is to report two Japanese sibs with MS and to review the diagnostic criteria and differential diagnosis.

CASE REPORTS

The first baby (case 1), a boy, was delivered at the 35th gestational week after a pregnancy which was apparently uneventful until there was obstructed labor due to enlarged after-coming head. No antenatal sonographic examination of the fetus had been performed. Birth weight was 2,300 g. Karyotype of the chromosomes was normal 46,XY. The mother was 25 years old at his birth and the father was 28 years old. The parents were not consanguineous. The infant died within several minutes after birth because of severe respiratory distress.

There was a huge occipital encephalocele, 25 cm in circumference, and a markedly protuberant abdomen (Fig. 1). Bilateral microphthalmia, low set ears, cleft palate, postaxial polydactyly of all four limbs and clubfeet were observed (Figs. 2, 3). The external genitalia was almost normal and not ambiguous. Polycystic kidneys (Potter II) and fibrosis of the portal areas and ductal proliferation and dilation of the liver were noted (Fig. 4).

The second baby, a girl, was delivered at the 40th gestational week and is alive, well, and normal.

The third baby (case 2), a boy, was delivered by prostaglandin termination at the 30th gestational week after sonographic determination of bilaterally enlarged polycystic kidneys. Birth weight was 1,930 g. Karyotype of the chromosomes was normal 46,XY. The infant died within several minutes after birth because of severe respiratory distress. The baby showed the triad of the condition (Fig. 5). Other than the triad, microphthalmia, low set ears, cleft palate and cleft lip, clubfeet, the patent ductus arteriosus, the patent foramen ovale, fibrosis of the portal areas, and ductal proliferation and dilation of the liver, and ambiguous external genitalia were observed.

DISCUSSION

Diagnostic Criteria

In spite of the span of over 170 years since the first report, and several extensive reviews on MS, the diagnostic criteria of MS is still somewhat unclear. It has been suggested that at least two of the three main manifestations, viz., occipital encephalocele, polycystic kidneys, and postaxial polydactyly, should be present to establish the diagnosis, or that two of these plus other frequently seen anomalies such as cleft lip and/or palate, microphthalmia, microcephaly, or small or ambiguous genitalia are required. Salonen reported, in 1984, a nationwide study on MS in Finland. In conclusion, she proposed that cystic dysplasia of the kidneys with fibrotic changes of the liver and occipital encephalocele or some other central nervous system (CNS) malformation were considered as minimal diagnostic criteria of MS.

Herriot et al. [1991] described three cases of MS in which the CNS anomaly was Dandy-Walker malformation (DWM). In their discussion, they provided an argument for the inclusion of DWM as one of the CNS

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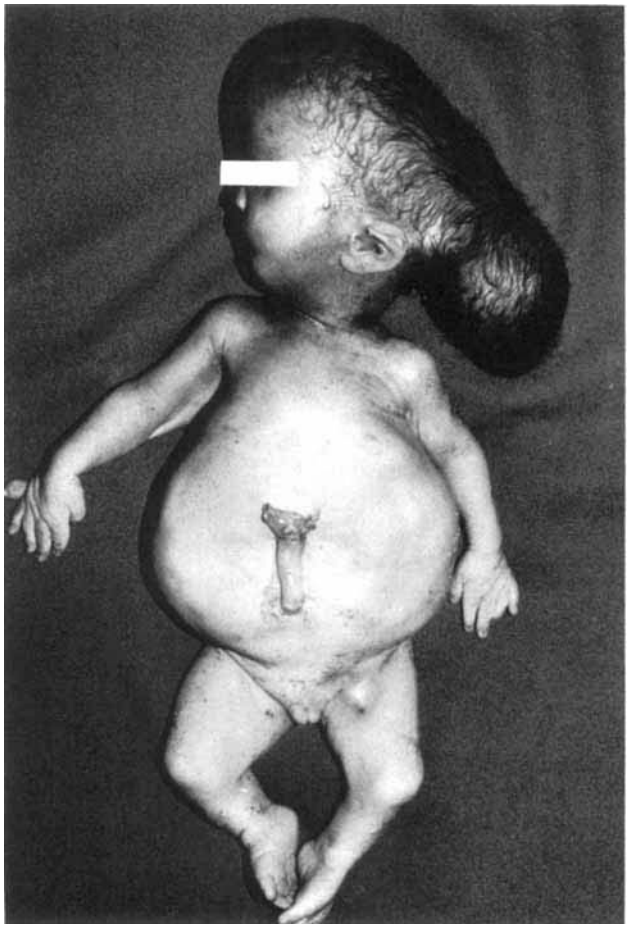

Fig. 1. External appearance of case 1 showing a huge occipital meningocele, a markedly swollen abdomen, postaxial polydactyly, and clubfeet.

Fig. 3. External appearance of both feet showing postaxial polydactyly and clubfeet.

malformations in MS. In the same year, Walpole et al. [1991] reported three sibs with DWM and cystic kidneys, one of whom also had hepatic fibrosis. They, however, suggested the possibility of a MS variant encompasses DWM but not polydactyly and encephalocele. Genuardi et al. [1993] noted that the presence of a DWM in a patient with renal cystic dysplasia, polydactyly, and hepatic fibrosis could be categorized as a case of cerebro-reno-digital syndrome. More recently, Summers and Donnenfeld [1995] described three sibs with varying manifestations of MS of whom two sibs were prenatally diagnosed with renal disease, polydactyly, and the DWM. They concluded that DWM should be included in the list of CNS abnormalities associated with MS. Recently, Paavola et al. [1995], in Finland, reported the locus for MS with multiple congenital anomalies mapped to chromosome 17q21–q24. The authors, to their regret, did not conduct a genome-wide study analyzing DNA samples from available Japanese families.

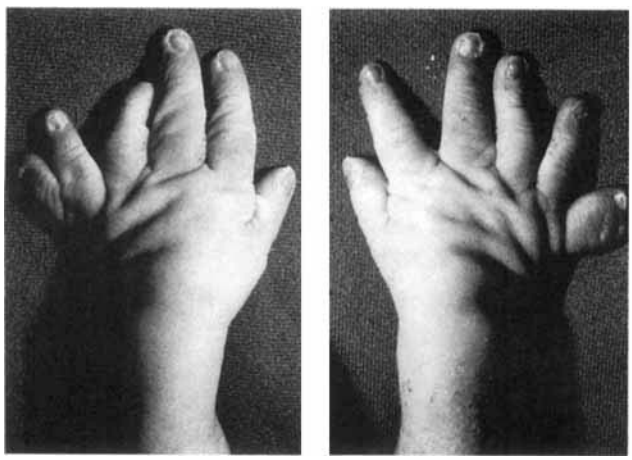

Fig. 2. External appearance of both hands showing postaxial polydactyly (heptadactyly).

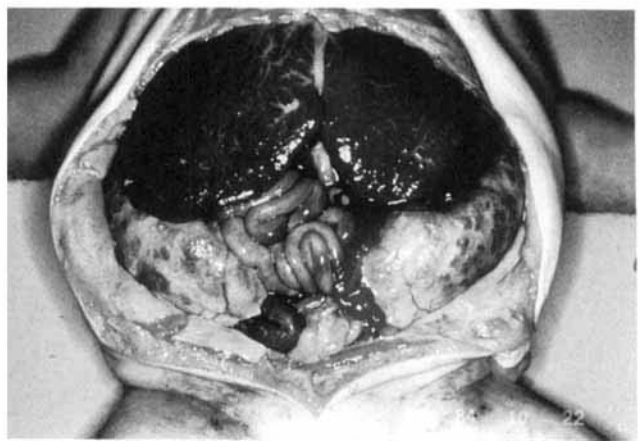

Fig. 4. Gross anatomical findings of abdominal viscera showing markedly swollen liver and polycystic kidneys.

Fig. 5. External appearance of case 2 showing an occipital meningocele, cleft lip, postaxial polydactyly, and clubfeet.

Differential Diagnosis

Marked similarities exist between MS and Patau syndrome or D_1-trisomy, except the normal karyotype and the occipital encephalocele. Short-rib polydactyly syndrome is a lethal disease but has neither cystic kidneys or encephalocele. The Ellis-van Creveld syndrome may show anomalies of CNS and urinary tract, especially DWM. MS, however, show no chondroectodermal dysplasia. Joubert syndrome, characterized by cerebellar vermis a/hypoplasia, developmental delay, hypotonia, abnormal eye movement, and abnormal breathing, should be included in the differential diagnosis since polydactyly, cystic kidneys, and hepatic fibrosis have occasionally been reported with this syndrome. In case 2 of the present report, a facies somewhat compatible with Potter syndrome was found. However, oligohydramnios was not observed.

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